

WHAT IS CLAIMED IS:

1. An isolated nucleic acid molecule comprising a polynucleotide having a nucleotide sequence selected from the group consisting of:

(a) a nucleotide sequence encoding a mutant of the PS1 polypeptide having the complete amino acid sequence in Figure 1 (SEQ ID NO:2);

(b) a nucleotide sequence encoding a mutant of the mature PS1 polypeptide having the amino acid sequence at positions 83-549 in Figure 1 (SEQ ID NO:2); and

(c) a nucleotide sequence complementary to any of the nucleotide sequences in (a) or (b).

2. The nucleic acid molecule of claim 1 wherein said polynucleotide has the complete nucleotide sequence in Figure 2 (SEQ ID NO:3).

3. The nucleic acid molecule of claim 1 wherein said polynucleotide has the nucleotide sequence in Figure 2 (SEQ ID NO:3) encoding the mutant PS1 polypeptide having the complete amino acid sequence in Figure 2 (SEQ ID NO:4).

4. The nucleic acid molecule of claim 1 wherein said polynucleotide has the nucleotide sequence in Figure 2 (SEQ ID NO:3) encoding the mutant of the mature PS1 polypeptide having the amino acid sequence at positions 83-549 in Figure 2 (SEQ ID NO:4).

5. An isolated nucleic acid molecule comprising a polynucleotide which hybridizes under stringent hybridization conditions to a polynucleotide having a nucleotide sequence identical to a nucleotide sequence in (a), (b) or (c) of claim 1.

6. An isolated nucleic acid molecule comprising a polynucleotide which encodes the amino acid sequence of an epitope-bearing portion of a mutant PS1 polypeptide having an amino acid sequence in (a) or (b) of claim 1.

7. A method for making a recombinant vector comprising inserting an isolated nucleic acid molecule of claim 1 into a vector.

8. A recombinant vector produced by the method of claim 11.

9. A method of making a recombinant host cell comprising introducing the recombinant vector of claim 8 into a host cell.

10. A recombinant host cell produced by the method of claim 13.

11. A recombinant method for producing a mutant PS1 polypeptide, comprising culturing the recombinant host cell of claim 10 under conditions such that said polypeptide is expressed and recovering said polypeptide.

12. An isolated mutant presenilin 1 (PS1) polypeptide comprising an amino acid sequence selected from the group consisting of:

- (a) the amino acid sequence shown in SEQ ID NO:4;
- (b) the amino acid sequence shown in SEQ ID NO:28;
- (c) the amino acid sequence shown in SEQ ID NO:30;
- (d) the amino acid sequence shown in SEQ ID NO:32; and
- (e) the amino acid sequence of an epitope-bearing portion of any one of the polypeptides of (a) - (d).

13. An isolated antibody that binds specifically to a mutant PS1 polypeptide of claim 12.

14. A method for diagnosing a patient having an increased likelihood of contracting Alzheimer's disease, comprising the steps of:

- a) obtaining from a patient a biological sample containing nucleic acid;
- b) incubating said nucleic acid with a probe which is capable of specifically hybridizing to a mutant PS1 gene under conditions and for time sufficient to allow hybridization to occur; and
- c) detecting the presence of hybridized probe, and thereby determining that said patient has an increased likelihood of contracting Alzheimer's disease.

15. A method for diagnosing a patient having an increased likelihood of contracting Alzheimer's disease, comprising the steps of:

- a) contacting a biological sample obtained from a patient with an antibody as claimed in claim 13 under conditions and for a time sufficient to allow binding of the antibody to the protein; and
- b) detecting the presence of the bound antibody.